



CHST3 gene

carbohydrate sulfotransferase 3

Normal Function

The *CHST3* gene provides instructions for making an enzyme called chondroitin 6-O-sulfotransferase 1 or C6ST-1. This enzyme has an important role in the development and maintenance of the skeleton. In particular, it is essential for the normal development of cartilage, which is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears.

The C6ST-1 enzyme modifies molecules called chondroitin sulfate proteoglycans, which are abundant in cartilage and give this tissue its rubbery, gel-like consistency. The C6ST-1 enzyme carries out a process known as sulfation, in which a chemical group called a sulfate is transferred from one chemical compound to another. Specifically, the enzyme takes sulfate from a molecule called 3'-phosphoadenyl-5'-phosphosulfate (PAPS) and adds it to a specific location on chondroitin sulfate proteoglycans. Sulfation of these molecules is a critical step in cartilage formation.

Health Conditions Related to Genetic Changes

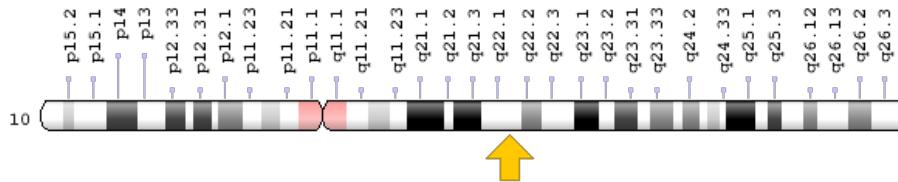
CHST3-related skeletal dysplasia

At least 24 mutations in the *CHST3* gene have been found to cause *CHST3*-related skeletal dysplasia, a condition characterized by progressive bone and joint abnormalities. Most of the mutations change single protein building blocks (amino acids) in the C6ST-1 enzyme. Other mutations result in the production of an abnormally short version of the enzyme. Each of these genetic changes reduces or eliminates the activity of C6ST-1, preventing it from transferring sulfate groups to chondroitin sulfate proteoglycans. Defective sulfation of these molecules disrupts the normal development of cartilage and bone, resulting in short stature, joint dislocations, and the other features of *CHST3*-related skeletal dysplasia.

Chromosomal Location

Cytogenetic Location: 10q22.1, which is the long (q) arm of chromosome 10 at position 22.1

Molecular Location: base pairs 71,964,362 to 72,013,564 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- C6ST
- C6ST-1
- C6ST1
- carbohydrate (chondroitin 6) sulfotransferase 3
- chondroitin 6-O-sulfotransferase 1
- CHST3_HUMAN
- galactose/N-acetylglucosamine/N-acetylglucosamine 6-O-sulfotransferase 0
- GST-0
- HSD

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Osteogenesis: The Development of Bones
<https://www.ncbi.nlm.nih.gov/books/NBK10056/>
- Essentials of Glycobiology (second edition, 2009): Proteoglycans and Sulfated Glycosaminoglycans
<https://www.ncbi.nlm.nih.gov/books/NBK1900/>

GeneReviews

- CHST3-Related Skeletal Dysplasia
<https://www.ncbi.nlm.nih.gov/books/NBK62112>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CHST3%5BTIAB%5D%29+OR+%28%28carbohydrate+sulfotransferase+3%5BTIAB%5D%29+OR+%28C6ST%5BTIAB%5D%29+OR+%28C6ST1%5BTIAB%5D%29+OR+%28C6ST-1%5BTIAB%5D%29+OR+%28chondroitin+6-O-sulfotransferase+1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- CARBOHYDRATE SULFOTRANSFERASE 3
<http://omim.org/entry/603799>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CHST3%5Bgene%5D>
- HGNC Gene Family: Sulfotransferases, membrane bound
<http://www.genenames.org/cgi-bin/genefamilies/set/763>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1971
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9469>
- UniProt
<http://www.uniprot.org/uniprot/Q7LGC8>

Sources for This Summary

- Hermanns P, Unger S, Rossi A, Perez-Aytes A, Cortina H, Bonafé L, Boccone L, Setzu V, Dutoit M, Sangiorgi L, Pecora F, Reicherter K, Nishimura G, Spranger J, Zabel B, Superti-Furga A. Congenital joint dislocations caused by carbohydrate sulfotransferase 3 deficiency in recessive Larsen syndrome and humero-spinal dysostosis. *Am J Hum Genet.* 2008 Jun;82(6):1368-74. doi: 10.1016/j.ajhg.2008.05.006. Erratum in: *Am J Hum Genet.* 2008 Aug;83(2):293.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18513679>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2427316/>
- Thiele H, Sakano M, Kitagawa H, Sugahara K, Rajab A, Höhne W, Ritter H, Leschik G, Nürnberg P, Mundlos S. Loss of chondroitin 6-O-sulfotransferase-1 function results in severe human chondrodysplasia with progressive spinal involvement. *Proc Natl Acad Sci U S A.* 2004 Jul 6; 101(27):10155-60. Epub 2004 Jun 23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15215498>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC454181/>
- Tsutsumi K, Shimakawa H, Kitagawa H, Sugahara K. Functional expression and genomic structure of human chondroitin 6-sulfotransferase. *FEBS Lett.* 1998 Dec 18;441(2):235-41.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/9883891>
- Tuysuz B, Mizumoto S, Sugahara K, Celebi A, Mundlos S, Turkmen S, Omani-type spondyloepiphyseal dysplasia with cardiac involvement caused by a missense mutation in CHST3. *Clin Genet.* 2009 Apr;75(4):375-83. doi: 10.1111/j.1399-0004.2009.01167.x.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19320654>
- Unger S, Lausch E, Rossi A, Mégarbané A, Sillence D, Alcausin M, Aytes A, Mendoza-Londono R, Nampoothiri S, Afroze B, Hall B, Lo IF, Lam ST, Hoefele J, Rost I, Wakeling E, Mangold E, Godbole K, Vatanavicharn N, Franco LM, Chandler K, Hollander S, Velten T, Reicherter K, Spranger J, Robertson S, Bonafé L, Zabel B, Superti-Furga A. Phenotypic features of carbohydrate sulfotransferase 3 (CHST3) deficiency in 24 patients: congenital dislocations and vertebral changes as principal diagnostic features. *Am J Med Genet A.* 2010 Oct;152A(10):2543-9. doi: 10.1002/ajmg.a.33641.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20830804>
- van Roij MH, Mizumoto S, Yamada S, Morgan T, Tan-Sindhunata MB, Meijers-Heijboer H, Verbeke JI, Markie D, Sugahara K, Robertson SP. Spondyloepiphyseal dysplasia, Omani type: further definition of the phenotype. *Am J Med Genet A.* 2008 Sep 15;146A(18):2376-84. doi: 10.1002/ajmg.a.32482.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18698629>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/CHST3>

Reviewed: October 2012

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services